Chair Nosse, Vice-Chairs Javadi and Nelson, and Members of the House Committee on Behavioral Health and Healthcare,

We write in support of House Bill 2741 because we believe that this policy would help other families avoid what happened to our son, Rockwell, after he was diagnosed through newborn blood screening. Our son suffered medical and developmental consequences that could have been prevented if we had been connected with the right resources from the start. We would like to share our story to illustrate the devastation that can result from families not having access to crucial resources after their child's diagnosis.

Our Son was born on May 15, 2012 at Mid Columbia Medical Center in The Dalles, Oregon. Rockwell was diagnosed with Phenylketonuria (PKU) at two weeks old in May of 2012 from a newborn screening. At this time we were living in Goldendale WA, a very tiny town north of The Dalles. When we got the call that our first born, tiny baby was diagnosed with an illness we had never heard of we felt so lost. We were told he needed a blood test asap. We scrambled to figure out where we needed to get him proper care. Mid Columbia Medical Center was not equipped to address his medical condition and referred us to Oregon Health and Science University (OHSU) for his treatment. We got an appointment at their earliest availability. This is where we were introduced to a physician as our PKU specialist. The physician diagnosed Rockwell with Persistent Hyperphenylalaninemia (HyperPhe). They said typically this type is very mild and no treatment is needed. As parents, hearing this of course gave us much relief.

Unfortunately, we were not given correct information on how often to test and went from monthly testing to annual testing. This was done at the advice of our physician. We were not made aware at the time that phenylalanine (Phe) levels can change drastically week to week. A fact that we learned years later but not from our original physician. We felt very alone in his diagnosis for the first several years. PKU is not common, we know no one with PKU and could not find a local community. Due to the physician's diagnosis of Rockwell's PKU being "so mild we don't need to do anything" also made us feel like outsiders in the PKU community. Like we were imposters. Our pediatrician was not even that knowledgeable in PKU and could not guide us or even see the signs that he was not getting the proper treatment.

By 2013, Rockwell was showing signs of developmental delays and we started him in Occupational Therapy and Speech Therapy. **We asked our physician multiple times if the issues he was experiencing could be related to his PKU. They would tell us that they were unaware of any possible connection and that it would be difficult to make that connection. They also told us that it would be hard for them to know because they had only been treating PKU patients for six years.** We asked the physician on more than one occasion if we needed to do anything since his levels were still high. They would always respond by telling us that we do not need to do anything because the levels are below 600 mg/dl. Not only is this absolutely incorrect in regard to his Phe levels, but there is no way of knowing if his levels were over 600 gm/dl throughout the year because he was only tested once per year instead of the recommended 52 times per year. This was during the time that we had the most difficulties with his developmental delays.

Rockwell has been diagnosed with Sensory Processing Disorder, Dysregulation, Receptive-Expressive Language Delay, and Attention Deficit Hyperactivity Disorder in addition to PKU. He is currently almost 13 years old and needed Occupational Therapy, Speech Therapy, Food Therapy as well as Special Education class with an Individualized Education Plan (IEP). We believe that Rockwell's subsequent diagnoses are directly related to the high levels of Phe that his brain was exposed to during the years that were not treated at the direction of the physician.

In addition to being misdiagnosed, Rockwell was not provided with the adequate medical supports that are suggested by the National PKU Alliance (NPKUA). The following information is from their guidelines for PKU treatment and diets:

- Regular clinical visits with a PKU care team are important to monitor and adjust diets as needed. This care team will consist of professionals trained to support individuals with PKU. This team should have the following members:
    - A Physician to oversee treatment, monitor labs and prescribe medications
    - A registered dietitian who helps plan nutritional choices for low protein consumption.
    - A genetic counselor to provide education regarding the genetics of PKU.
    - A social worker to help facilitate communication between the team and the parents and patient.
    - Additionally, some clinics also provide a psychologist to perform neurological assessments of the child.

Rockwell was not given a team but solely treated by the physician. We were never introduced or offered a dietitian. A dietician would have known that baby formula is higher in protein than breastmilk and would have offered medical baby formula and food. The physician knew Rockwell was on infant formula due to my low milk production. We did not have regular office visits and our visits went from monthly to annually. His Phe levels were tested 3 times in his first year of life (2012), twice in 2013, then once a year until 2019. As previously stated, Phe levels change weekly and can cause long lasting damage to the brain if not monitored and treated. We were never told or made aware that OHSU has a PKU team or another physician that is an actual PKU specialist until October of 2019. As soon as we were made aware, we requested his soonest available appointment. On January 28th, 2020, we met with this PKU specialist for the first time. We explained all our issues with the lack of treatment that Rockwell had received from May 2012 to December 2018, his disabilities and struggles, as well as our own findings as we constantly researched ourselves. This PKU specialist was upset to hear the issues we had experienced with our previous physician. They confirmed to us that Rockwell's disabilities are absolutely from his PKU levels. If this PKU specialist would have been seeing Rockwell since birth and was made aware of the difficulties he was experiencing, they would have acted on the information by starting a treatment plan. Additionally, this PKU specialist would have had Rockwell evaluated by a psychologist as soon as he was made aware of his developmental difficulties as stated in the guidelines from the NPKUA. The PKU specialist was very apologetic for the suffering Rockwell has had to go through as well as ours as his parents.

At our first appointment with our new team, on December 18th, 2018, we were informed that new studies have shown that Phe levels need to be below 360 mg/dl. This was very shocking to us, as Rockwell's levels had been 400-600+mg/dl since birth. Once we returned home after receiving this news, we decided to do some of our own research. We asked other members of community groups on social media when they were informed of this important change and were extremely upset when we finally found out that the therapeutic range below 360 mg/dl is not new news at all. We went back through Rockwell's labs since birth and each result says, "Phenylalanine above the desired therapeutic range. The desired therapeutic range is 120-360mg/dl." This led us to conduct further research to find out what care Rockwell should have received, what his diagnosis actually is, the damage that had been done to his brain, and how we can help him moving forward. We had difficulty even finding this information because we were referring to our son's condition as HyperPhe, but as we now know his correct term is PKU. We only found the information about his condition not being referred to as HyperPhe because a PKU community member directed us to resources on PKU guidelines and levels on October 8, 2019. This is what led us to find the guidelines outlined by the NPKUA. On November 4, 2019, we received guidelines from NPKUA which included a 2014 update to guidelines for treatment of PKU, and a 2017 paper about Moderate/ Mild PKU, additionally documentation suggests not referring to the severity of PKU as the diagnosis, i.e. HyperPhe or Mild PKU, because levels can change substantially. With this additional information we discovered these guidelines mostly revolve around brain function and child development. Our current PKU specialist is on the NPKUA Scientific-Advisory-Board and the department had access and was aware of these guidelines. If we would have known earlier that we should have been researching PKU and not HyperPhe, we would have been able to adequately research our son's condition and advocate for him when we were misled by the first PKU specialist we saw. We should have been made aware of these guidelines by OHSU and received proper care long before having to do our own research, and finally getting the correct treatment after our son was six years old.

Since his new care started in 2019, we test Rockwell's Phe levels every single week. He started the medication Kuvan, which has shown big improvements in his cognitive abilities. We talk to a clinic dietitian at every visit. We monitor his protein intake and have been given medical formula and food to try. Rockwell's disabilities affect his willingness to try different foods, so starting him on a PKU friendly diet as well as medical food and formula was difficult. All of this should have been started from birth.

Rockwell has suffered greatly due to years of misinformation and incorrect treatment. He was not able to handle being around other children or adults. It would cause him severe anxiety, over stimulation and dysregulation. Due to the associated complications from years of high Phe levels, Rockwell was not socialized until he was five years old, attending preschool in a special needs classroom with an IEP. Rockwell could not attend birthday parties, family functions, have a babysitter, play at the park, or go to restaurants and stores. Even having his grandparents over would cause extreme stress. We would have to restrain him from hitting his head on walls or the floor when he got so upset. Rockwell could not speak clearly until he was five years old and still struggles with speech today. It is all very isolating for us all. Rockwell's best friend was his

Occupational Therapist, who he has had since he was two and a half years old. He still struggles with socializing with his peers. Everyday tasks are difficult and upsetting. Routine dental checkups and procedures required sedation leaving us with large dental bills. Routine doctor appointments were so hard because he would not let any doctor touch him. This led to going years without being properly examined as he grew. Every appointment was filled with yelling, crying, and having to physically restrain Rockwell to prevent him from hurting himself. Milestones were delayed. Experiences with our first-born son were missed. Something as simple as reading him books was not possible till he was five years old. Rockwell's early childhood was traumatizing to him and us. Having to relive everything we went through brings us great anxiety, stress, sadness and anger. Knowing that he did not have to suffer, that this was preventable affects us every day.

Having someone follow up with us in the years after his diagnosis could have helped guide us and potentially changed Rockwells health greatly. It would have helped us feel more in control of the situation instead of feeling lost, scared and stressed. Having someone hear our concerns connect us with current requirements for levels and treatments would have let us know right away that he was not receiving the correct treatment. Just having someone listen to us about our concerns about his current treatment with potential to help would have given us hope. Living in a very rural town at the time also hindered us from being able to have access to more interventions, but if we had an advocate to help guide us then we could have known what we needed to do. Receiving information from the Oregon Health Authority about organizations like PKU Northwest, would have connected us to a community that knows what we are going through instead of us searching for online forums on our own.

Rockwell is very intelligent. It pains us so much knowing that his potential was hindered by misdiagnosis and mistreatment. Our family will be living with the lifelong consequences from this misdiagnosis. Any outside intervention during years of stress and confusion could have changed the trajectory of our son’s life. We support House Bill 2741 because we do not want this to happen to families in Oregon.

Thank you,
Seth and Katie Potts